



## Interaction of CYP46A1 with CFH, LOC387715 and HTRA1 gene polymorphisms in age-related macular degeneration

Brice Dugas, Cynthia Fourgeux, Bénédicte Buteau, Lucy Martine, Ingemar Bjorkhem, Niyazi Acar, Alain M. Bron, Frédéric Nicot, Catherine P. Creuzot Garcher, Lionel Brétillon

### ► To cite this version:

Brice Dugas, Cynthia Fourgeux, Bénédicte Buteau, Lucy Martine, Ingemar Bjorkhem, et al.. Interaction of CYP46A1 with CFH, LOC387715 and HTRA1 gene polymorphisms in age-related macular degeneration. Association for Research in Vision and Ophthalmology annual congress, May 2009, Fort Lauderdale, Florida, United States. 1 p. hal-01136998

**HAL Id: hal-01136998**

**<https://hal.science/hal-01136998>**

Submitted on 30 Mar 2015

**HAL** is a multi-disciplinary open access archive for the deposit and dissemination of scientific research documents, whether they are published or not. The documents may come from teaching and research institutions in France or abroad, or from public or private research centers.

L'archive ouverte pluridisciplinaire **HAL**, est destinée au dépôt et à la diffusion de documents scientifiques de niveau recherche, publiés ou non, émanant des établissements d'enseignement et de recherche français ou étrangers, des laboratoires publics ou privés.

## Interaction of CYP46A1 with CFH, LOC387715 and HTRA1 Gene Polymorphisms in Age-Related Macular Degeneration

B. Dugas, Jr.<sup>1</sup>, C. Fourgeux, Jr.<sup>2</sup>, B. Buteau, Jr.<sup>2</sup>, L. Martine, Jr.<sup>2</sup>, I. Björkhem, Sr.<sup>3</sup>, N. Acar, Sr.<sup>2</sup>, A. Bron, Sr.<sup>1</sup>, F. Nicot, Jr.<sup>1</sup>, C. Creuzot-Garcher, Sr.<sup>1</sup> and L. Bretillon, Sr.<sup>2</sup>

<sup>1</sup>Ophthalmologie, CHU Dijon, Dijon, France

<sup>2</sup>Umr1129 flavic, inra, Eye and Nutrition Research Group, Dijon, France

<sup>3</sup>Karolinska University Hospital Huddinge, Department of Laboratory Medicine, Stockholm, Sweden

**Commercial Relationships:** B. Dugas, Jr., None; C. Fourgeux, Jr., None; B. Buteau, Jr., None; L. Martine, Jr., None; I. Björkhem, Sr., None; N. Acar, Sr., None; A. Bron, Sr., None; F. Nicot, Jr., None; C. Creuzot-Garcher, Sr., None; L. Bretillon, Sr., None.

**Support:** None.

**Clinical Trial:** www.clinicaltrials.gov NCT00629044

### Abstract

**Purpose:** To estimate the association and interaction of single nucleotide polymorphism (SNP) in cholesterol-24S-hydroxylase (CYP46A1) gene with HTRA1, LOC387715, CFH SNPs with age-related macular degeneration (AMD) in a North-East French population.

**Methods:** Cross-sectional study involving 142 AMD patients with exudative AMD or geographic atrophy and 70 unrelated control subjects. SNPs were genotyped in CYP46A1, HTRA1, LOC387715, and CFH genes. Plasma 24S-hydroxycholesterol, the metabolic product of CYP46A1, was quantified. Sex, age, alleles, and genotype frequencies between AMD patients and controls were compared using the  $\chi^2$  and Student t-tests. Odd-ratios (OR) and 95% confidence intervals (CI) were calculated by logistic regression to assess the relative association between disease and age, sex, and genotypes.

**Results:** The SNP rs754203 in CYP46A1 was not associated with AMD (OR=1.1, 95% CI=0.78-1.43, p=0.76). The OR for risk of AMD was 2.1 (95% CI=1.1-4.4, p=0.03) for the A-allele of rs11200638 in HTRA1, 2.8 (95% CI=1.4-5.5, p=0.002) for the T-allele of rs10490924 in LOC387715, and 1.5 (95% CI=1.1-2.0, p=0.004) for the C-allele for rs1061170 in CFH. These associations were found only in patients with exudative AMD but not with geographic atrophy. An OR of 11.3 (95% CI=0.7-170, p=0.003) was obtained for carriers with both CC-genotype in CFH and TT in CYP46A1.

**Conclusions:** The TT-genotype of rs754203 in CYP46A1 conferred a higher risk for exudative AMD in patients who carry the CC-genotype of rs1061170 in CFH.

**Keywords:** age-related macular degeneration • choroid: neovascularization • genetics